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INCIDENCE OF PAPILLON-LEFÈVRE SYNDROME IN A KASHMIRI POPULATION

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ABSTRACT

Papillon-Lefèvre syndrome is an autosomal recessive disorder with a rare occurrence. The characteristic features of this rare syndrome are palmarplantar hyperkeratosis and early onset of a severe destructive periodontitis. There is a diverse etiopathology associated with the syndrome; but a recent report has shown the syndrome is due to mutations of the cathepsin C gene. A detailed analysis was undertaken to evaluate the prevalence of this rare disorder in a kashmiri population using clinical, laboratory and radiographic investigations.

KEYWORDS: Hyperkeratosis, Papillon-Lefèvre Syndrome & Periodontitis

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Original Article

INTRODUCTION

The characteristic features of Papillon–Lefèvre syndrome (PLS) are hyperkeratosis of hands and feet and generalized aggressive periodontitis in both the primary and the permanentdentition.[1] PLS is a rare disorder with auto somal recessive inheritance. consanguinity of parents is seen in 20% to 40% of the cases.[1]Associated clinical features include calcification of the falxcerebri and the choroid plexus.[2-4] Altered immunity to fight infections is seen in 20–25% of such patients [1,3,5,6] The recent diagnostic methods revealed the genetic defectin PLS has been mapped to chromosome 11q14–q21, which involves mutations of cathepsin C.[7,8] Studies in PLS patients display 90% reduction in cathepsin C activity.[8,9] The cathepsin C gene encodes a cysteine-lysosomalprotease also known as *dipeptidyl-peptidase*. The expression of cathepsin-C is mainly seen in the epithelial regionssuch as palms, soles, knees, and keratinized oralgingiva. These areas aremostlyaffected by PLS. The etiopathology of periodontal involvement remains unknown. Periodontitis occurs after tootheruption(10-17). There is huge Plaque accumulation in thedeep crevices and halitosis canoccur. Theprimary incisors are the first teeth to show periodontitis. The whole primary dentition is shedded by the age of four or five years.[1,4] Routine periodontal treatmentwith scaling, and rootplanning and other treatment modalities have proven to be of no help. [18-25].

There has not yet been any studies/case reports of this rare syndrome in Kashmiri population. The aim of this study was therefore to evaluate the frequency of Papillon-Lefèvre syndrome in a Kashmiri population using clinical, laboratory and radiological investigations.

MATERIAL AND METHODS

The study was conducted jointly in the departments of periodontics and conservative dentistry and

endodontics government dental college srinagar. A total of 10000 patients (6000 males and 4000 females) attending the general opd were included in the study. A detailed procedure was followed to evaluate each patient.

General and Extra-Oral Examination

Extra-oral examination of each patient was done to evaluate the presence of any yellowish, keratotic, confluent plaques affecting the skin ofpalms and soles. Examination ofnails and hair were also done for each patient. Extra-oral examination of two cases revealed symmetric, well-demarcated, yellowish, keratotic, confluent plaques affecting the skin ofpalms and soles and extending onto the dorsal surfaces. Their nails were discolored, but hair was normal. Well circumscribed, psoriasiform, erythematous, scaly plaques werepresent on the knees bilaterally [Figures 1 and 2].

Intraoral examination

Intraoral examination of each case was done to evaluate any mobile and missing teeth, presence of periodontal pockets or presence of agreesive periodontitis. One case revealed missing upper left permanent centralincisors, lower left permanent central and lateral incisors Severe gingival inflammation, abscess formation, and deep periodontal pockets were noticed. Severe mobility affecting all the teeth, with heavy deposits of plaque andcalculus and halitosis were also present [Figure 3].Intraoral examination of case 2 showed deep periodontalpockets, gingival inflammation, and mobility of lower anteriorteeth [Figure 4]. All primary teeth were exfoliated and no toothwas missing.

Radiographic Findings

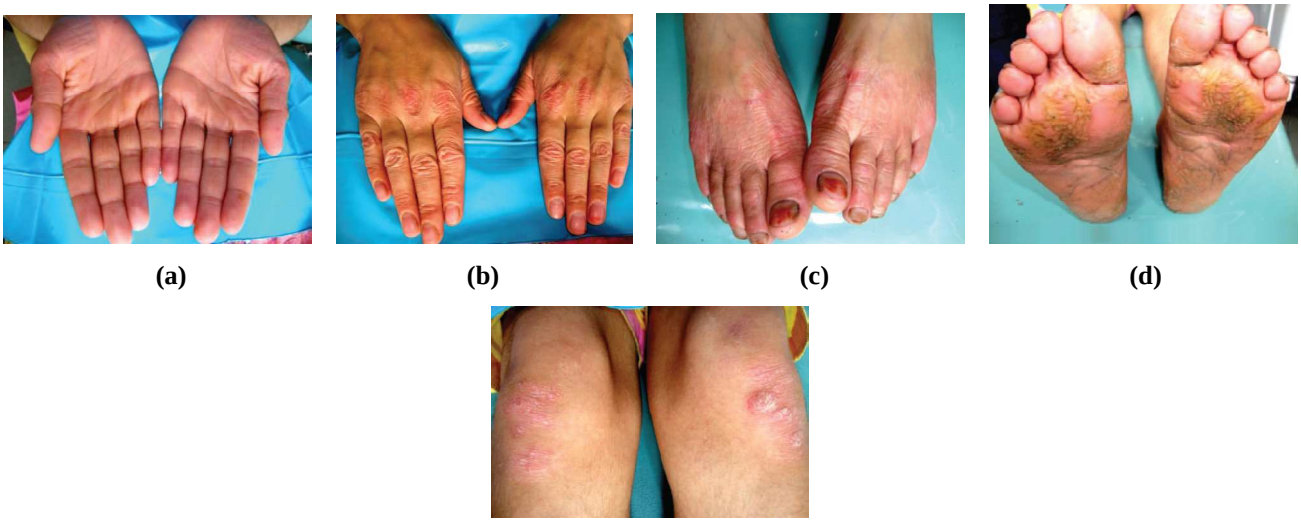
Orthopantomographic examination of each case was done to evaluate the destruction of alveolar bone. Case 1 showed severegeneralized destruction of alveolar bone. The mandibular right first molar was almost entirely out of its socket and was almostwithout bone support [Figure 5]. Case 2 showed generalizedloss of alveolar bone [Figure 6]. On lateral skull radiograph, there was no evidence ofintracranial calcification in both the patients [Figure 7].

Laboratory Investigation

Laboratory investigation was carried out, which includedhematological and biochemical assessment. The results were within normal limits.

(a) **(b)** **(c)** **(d)**

Figure 1: (a) Case 1 Presenting with Yellowish, Keratotic, Confluent Plaques Affecting the Skin of Palmar Surfaces of Hands; (b) Case 1 Presenting with Keratotic Plaques on the Dorsal Surfaces of Hands; (c) Keratotic Plaques Affecting the Dorsal Surface of Feet; (d) Several Confluent Plaques on Soles



(a) **(b)** **(c)** **(d)**

(e)

Figure 2: (a) Case 2 Presenting with Keratotic, Confluent Plaques Affecting the Skin of Palmar Surfaces of Hands; Keratotic Plaques Affecting the Dorsal Surfaces of Hands; (c)Plaques Affecting the Dorsal Surfaces of Feet; (d) Several Confluent Plaques Affecting the Soles; (e) In Case 2, well-Circumscribed, Erythematous, Scaly Plaques on the Knees bilaterally are also Noticed

Figure 3: Intraoral Examination of Case 1 Showing Missing Upper Left Permanent

Central incisor, lower left permanent central and lateral incisors and permanent right

Lower central incisor. Severe gingival inflammation, abscess formation, and deep

Periodontal pockets were noticed. Severe mobility affecting all the teeth, with heavy

Deposits of plaque and calculus and halitosis, were also present

Figure 4: Intraoral Examination of Case 2 Showing Deep Periodontal Pockets, Gingival Inflammation, and Mobility of Lower Anterior Teeth

Figure 5: OPG of Case 1 Showing Severe Generalized Destruction of Alveolar Bone

The mandibular right first molar was almost entirely out of its socket with not much bone support

Figure 6: OPG of Case 2 Showing Generalized Destruction of Alveolar Bone

See the severe periodontal destruction in upper right first molar

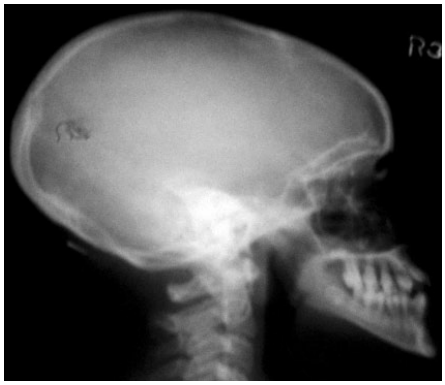


Figure 7: Lateral Skull View of Case 1 Showing no Evidence of Intracranial Calcification

RESULTS

In view of the above findings, two cases were diagnosed as PLS out of 10, 000 patients with an overall percentage of 0.01% in the study population.

DISCUSSIONS

PLS has shown profound influence on the psychological and social life of the affected children. The aim of the dental clinician in such cases should be thorough dental examination andcounselingof the patients parents. The etiopathology of this rare syndrome is not known some studies have proposed immunologic, microbiologic, and genetic bases. *A. actinomycetemcomitans* has been shown to be the main microorganism responsible for the aggressive periodontitis in these patients.[5,12,26,27,28] the current research has revealed the inactivation of the cathepsin Cgene in

these patients.[29] In our study dermatological, periodontal, and radiological features strongly suggested the diagnosis of PLS. Consanguineous marriage of the parents of the affected children has been reported for this syndrome, [29] A definite treatment plan has not been yetdevised for such patients;conventional periodontaltherapy, oral hygiene instructions, and systemic antibiotics can be employed.[27]Further research is needed for evaluation and treatment of such children. Stem cell therapy can beemployed in the future for the dental treatment ofsuch children.

CONCLUSIONS

PLS threatens children and their parents with the prospect of edentulism if left untreated. Hence, early diagnosisand intervention is essential. For edentulous patients, oralrehabilitation is required; which includes partial or completedenture prosthetic replacement (according to the age of thepatient). Osseointegrated implants are an option for the futureand can have a great impact psychosocially by restoringesthetics as well as function. The periodontist is the first member of the health team to seeand treat the patients afflicted with such unusual syndromessuch as PLS, and therefore, greater awareness of this syndromewill be helpful in identifying more cases for further study.

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